

**What is claimed:**

1. An isolated polynucleotide comprising the nucleic acid sequence selected from the group consisting of:
  - (a) the polynucleotide sequence of SEQ ID NO: 1,
  - 5 (b) the polynucleotide sequence of SEQ ID NO: 3.
  - (c) the polynucleotide sequence of SEQ ID NO: 14;
  - (d) the polynucleotide sequence of SEQ ID NO: 16;
  - (e) a polynucleotide that hybridizes under the following stringent conditions to the complement of any one of (a)-(e);
    - 10 (1) hybridization at 65°C in a hybridization buffer comprising 0.5 M NaHPO<sub>4</sub>, and
    - (2) washing at 65°C in a wash solution comprising 1x SSC.
2. An isolated polypeptide encoded by the polynucleotide of claim 1.
- 15 3. An isolated polypeptide of claim 2, wherein the polypeptide comprises the amino acid sequence of SEQ ID NO: 2.
4. An isolated polypeptide of claim 2, wherein the polypeptide comprises 20 the amino acid sequence of SEQ ID NO: 15.
5. An antibody that specifically bind a polypeptide of claim 2.
6. A composition comprising the polynucleotide of claim 1 and a carrier.
- 25 7. A composition comprising the polypeptide of claim 2 and a carrier.
8. A method of detecting the Hydin gene comprising steps of:
  - (a) contacting a biological sample with a compound that binds to the 30 polynucleotide of claim 1; and
  - (b) detecting binding between the compound and the polynucleotide, wherein binding indicates the presence of the Hydin gene in the sample.

9. A method of detecting the Hydin polypeptide comprising steps of:
    - (a) contacting a biological sample with a compound that binds to the polypeptide encoded by the polynucleotide of claim 1; and
    - (b) detecting binding between the compound and the polypeptide,
  - 5 wherein binding indicates the presence of the Hydin polypeptide in the sample.
10. The method of claim 9, wherein the compound that binds the polypeptide is an antibody,
  - 10 11. A method of detecting a mutation in the human Hydin gene comprising steps of:
    - (a) contacting a biological sample with a compound that binds to the polynucleotide having the nucleic acid sequence of SEQ ID NO: 14; and
    - (b) detecting binding between the compound and the polynucleotide,
  - 15 wherein binding indicates the presence of a mutation in the human Hydin gene in the sample.
12. The method of claim 11, wherein the mutation is located at a position
  - 20 that corresponds to the position of the OVE459 mutation within the murine Hydin gene.
13. A method of diagnosing hydrocephalus in a human comprising detecting a mutation in the Hydin gene according to the method of claim 11; wherein
  - 25 the presence of the mutation in the human Hydin gene indicates a probability of the human developing hydrocephalus.
14. The method of claim 13 , wherein the mutation is located at a position
  - that corresponds to the position of the OVE459 mutation within the murine Hydin
  - 30 gene.
15. The method of claim 14, wherein the mutation is detected in a prenatal human.

16. A method of diagnosing a cilia dysfunction-related disorder comprising detecting a mutation in the Hydin gene according to the method of claim 11; wherein the presence of the mutation in the Hydin gene indicates a probability of the developing a ciliary dysfunction-related disorder.

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17. The method of claim 16, wherein the cilia related disorder is selected from the group consisting of Kartagerner syndrome, primary cilia dyskinesia, chronic sinusitis, male infertility, deafness or kidney failure.